



## APTX gene

aprataxin

### Normal Function

The *APTX* gene provides instructions for making a protein called aprataxin that is involved in the repair of damaged DNA or RNA, the chemical cousin of DNA. Aprataxin is located in the nucleus of cells and is produced in various tissues, including the brain, spinal cord, and muscles. Different regions of the aprataxin protein aid in its DNA or RNA repair function by allowing the protein to interact with other DNA repair proteins and to attach (bind) to DNA or RNA molecules. At the site of damage, aprataxin modifies the broken ends of the DNA or RNA strands so they can be joined back together.

### Health Conditions Related to Genetic Changes

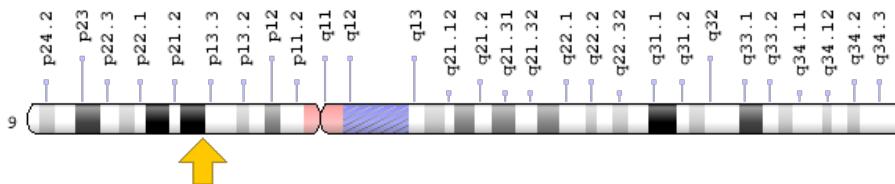
#### ataxia with oculomotor apraxia

At least 30 mutations in the *APTX* gene have been found to cause ataxia with oculomotor apraxia type 1. This condition is characterized by difficulty coordinating movements (ataxia) and problems with side-to-side movements of the eyes (oculomotor apraxia). Most mutations change single protein building blocks (amino acids) in aprataxin, resulting in an unstable aprataxin protein that is quickly broken down in the cell. A lack of functional aprataxin disrupts DNA repair and can lead to an accumulation of DNA damage in cells, particularly affecting brain cells in the part of the brain involved in coordinating movements (the cerebellum). This accumulation can lead to cell death in the cerebellum, causing the characteristic movement problems of ataxia with oculomotor apraxia type 1.

## Chromosomal Location

Cytogenetic Location: 9p21.1, which is the short (p) arm of chromosome 9 at position 21.1

Molecular Location: base pairs 32,883,872 to 33,025,129 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AOA
- AOA1
- APTX\_HUMAN
- ataxia 1, early onset with hypoalbuminemia
- AXA1
- EAOH
- EOAHA
- FHA-HIT

## Additional Information & Resources

### Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): DNA Repair  
<https://www.ncbi.nlm.nih.gov/books/NBK9900/>

### GeneReviews

- Ataxia with Oculomotor Apraxia Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1456>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28APTX%5BTIAB%5D%29+OR+%28aprataxin%5BTIAB%5D%29%29+OR+%28AOA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

## OMIM

- APRATAXIN  
<http://omim.org/entry/606350>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_APTX.html](http://atlasgeneticsoncology.org/Genes/GC_APTX.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=APTX%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=15984](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15984)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/54840>
- UniProt  
<http://www.uniprot.org/uniprot/Q7Z2E3>

## **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/APTX>

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